Joint Hypermobility

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Joint Hypermobility Syndrome

- Increased range of motion beyond its normal range *(recognizing that range of motion is on a continuum)*
- 10% general population *(?)*
- *Not clear if Joint Hypermobility Syndrome and Ehlers Danlos Hypermobility type are the same*
- Can be associated with musculoskeletal pain
  - Easier/more frequent joint dislocations
  - Probable early degenerative arthritis
  - May also have muscular pain
- Important to recognize
  - Reassurance to patient - no evidence for more serious disease *(i.e. cancer)*
  - Give a name/reason to the patient for what they are feeling – *allows them to learn about it and develop a coping strategy*
  - Prevent unnecessary laboratory tests or X-rays
  - Rarely, identify a more serious cause of joint hypermobility

Pathophysiology

- Unclear
- Common Hypotheses:
  - Biomechanical
    - Localized biomechanical overload during activity
    - Joint *(tendon, ligament)* instability → microtrauma, compensation of movement patterns leading to overload in other MSK areas
    - Proprioception altered
  - Deconditioning
  - Generalized hyperalgesia
    - Up-regulation of central pain processes
    - Decreased responsiveness to lidocaine and opioids

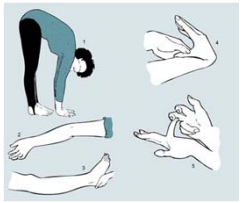
Screening Questions

- Questionnaire
  - Can you now *(or could you ever)* place your hands flat on the floor without bending your knees?
  - Can you now *(or could you ever)* bend your thumb to touch your forearm?
  - As a child, did you amuse your friends by contorting your body into strange shapes or could you do the splits?
  - As a child or teenager, did your kneecap or shoulder dislocate on more than 1 occasion?
  - Do you consider yourself 'double-jointed'?
- ≥ 2 are positive, Sensitivity 84%, Specificity 80%

Hakim AJ, Grahame R. A simple questionnaire to detect hypermobility: an adjunct to the assessment of patients with diffuse musculoskeletal pain. Int J Clin Pract 2003;57(3):163–6.

Diagnosis

- Beighton Score
  - Place hands flat on floor *(1)*
  - Hyperextend elbow > 10° *(2)*
  - Hyperextend knee > 10° *(3)*
  - Bend thumb to forearm *(4)*
  - Dorsiflex pinkie > 90° *(5)*
- 1 point for each *(bilateral except touching floor)*
- 9 points possible



Brighton Criteria (1998)

- Major
  - Beighton score of ≥ 4
  - Arthralgia for ≥ 3 months in ≥ 4 joints
- Minor
  - Beighton score of < 4
  - Arthralgia in < 4 joints; back pain/spondylolysis/listhesis
  - ≥ 3 soft tissue problems (epicondylitis, bursitis, tenosynovitis)
  - Marfanoid habitus (tall and slim)
  - Skin: striae, hyperextensibility, thin skin, abnl scarring
  - Eye: drooping eyelids, myopia, antimongoloid slant
  - Varicose veins, hernia or uterine/rectal prolapse
- Diagnosis
  - 2 major *-or-*
    - 1 major and 2 minor *–or-*
    - 4 minor *–or-*
    - 2 minor if affected 1st-degree relative
  - Exclude Marfan's and EDS *(other than hypermobility type)*
- Caveat: Criteria designed for epidemiology research
  - Gives organized approach to pt

Grahame R, Bird HA, Child A, et al. J Rheumatology 2000; 27(7): 1777-9.

Differential Diagnosis

- EDS classic types
- Marfan's Syndrome
- Osteogenesis Imperfecta
- Others
  - Loeys-Dietz
  - Arterial Tortuosity Syndrome
  - Lateral Meningocele Syndrome

Ehler's Danlos Types

- Classic: Skin hyperextensibility w widened atrophic scars and joint hypermobility
- Vascular: Arterial, intestinal, uterine fragility with (catastrophic) rupture
- Kyphoscoliotic: Scoliosis at birth w scleral fragility with joint hypermobility
- Arthrochalasia: Recurrent, severe joint subluxations w severe joint hypermobility
- Dermatosparaxis: Severe skin fragility

Ehler's-Danlos Hypermobility Type (III)

- Joint Hypermobility
- Soft, thin stretchy skin with thin scars
- Family history may be present
- Systemic involvement
  - **Musculoskeletal pain** (often labeled fibromyalgia)
    - Nonanatomic
    - Post-exertional worsening
  - **Autonomic Dysfunction**
    - Postural tachycardia syndrome (POTS) – most common
  - **Gastrointestinal dysmotility**
    - Constipation
    - Rectoceles
    - GI overlap with GU
    - Nausea, abdominal pain, reflux, bloating

Osteogenesis Imperfecta

- Joint hypermobility
- Susceptibility to bone fractures
- Blue sclera
- Opalescent dentine
- Sensorineural deafness

Marfan's Syndrome

- Hereditary connective tissue disease
- Autosomal dominant
- FBN1 gene mutations (encoding fibrillin 1 protein)
- Manifestations
  - Cardiovascular – aortic dilatation or aneurysm
  - Skeletal – joint hypermobility + Marfanoid Habitus
  - Ocular – ectopia lentis
  - Others
- Marfanoid Habitus:
  - Tall, thin
  - Arachnodactyly
  - Dolichostenomelia: arm span: height ratio ≥ 1.03

Marfan's Syndrome (MFS):
Diagnostic Criteria

- (+) Family History
  - Ectopia Lentis (EL) = MFS
  - Systemic ≥ 7 = MFS
  - Aortic diameter (Ao)
    - > 20 years, Z ≥ 2 = MFS
    - < 20 years, Z ≥ 3 = MFS
- (-) Family History
  - Ao (Z ≥ 2) + EL = MFS
  - Ao (Z ≥ 2) + FBN1 = MFS
  - Ao (Z ≥ 2) + Systemic (≥ 7) = MFS
  - EL + FBN1 = MFS

- *Z age<40 years = [measured aortic root diameter – (0.97 + 1.12 × BSA)]/0.24*
- *Z age>40 years = [measured aortic root diameter –(1.92 + 0.74 × BSA)]/0.37*

Scoring of Systemic Features

- Thumb and wrist sign – 3
- Wrist or thumb sign – 1
- Pectus carinatum – 2
- Hind foot deformity – 2
- Pneumothorax – 2
- Dural ectasia – 2
- Protrusio acetabuli – 2
- Reduced upper segment/lower segment and increased arm/length ratio and no severe scoliosis – 1
- Scoliosis or thoracolumbar kyphosis – 1
- Reduced elbow extension – 1
- Facial features (3/5) – 1 (dolichocephaly, enophthalmos, down-slanting palpebral fissures, malar hypoplasia, retrognathia)
- Skin striae – 1
- Myopia > 3 diopters – 1
- Mitral valve prolapse – 1
- Pectus excavatum or chest asymmetry – 1
- Plain pes planus – 1

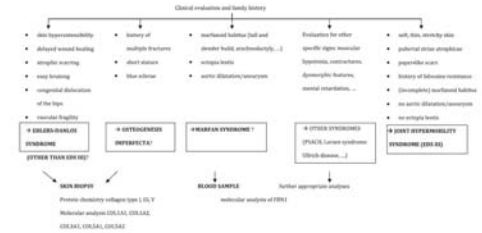
Back

Others:

Uncommon to Rare

- Loeys-Dietz: autosomal dominant
  - aortic aneurysm
  - Hypertelorism
  - Bifid uvula or cleft palate
- Arterial Tortuosity Syndrome: Autosomal dominant
  - Tortuosity and elongation of large/medium sized arteries
  - Tendency for aneurysm formation
- Lateral meningocele Syndrome
  - Widespread spinal lateral meningoceles protruding through intervertebral spaces

Approach to a patient



From: Fikree A, Aziz Q, Grahame R. Joint Hypermobility Syndrome. Rheum Dis Clin NA 2013; 39, 419-430 attributed to Malfait F, et al. Rheumatology (Oxford) 2006;45(5): 502-7.

Therapy

- Physical Therapy
- Avoidance of activity causing dislocations
- Occupational therapy (if finger laxity present)
- Cognitive Behavioral Therapy
- Family history, suspected other disease (e.g. Marfan's, Osteogenesis Imperfecta) – referral to pediatrics genetics clinic.

Take Home Points

- Joint mobility is a continuum
- Joint hypermobility syndrome: chronic joint pain (and more)
- Diagnose with Beighton and Brighton scores
- Look for other causes
  - EDS
  - Marfan's
  - Osteogenesis Imperfecta
- Therapy: Physical therapy (and others)
- Refer if suspicion for other causes is high

Questions?

- References

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